

**WE CLAIM:**

1. An isolated nucleic acid molecule encoding a protein with a RING-  
5 finger domain and 6 NHL-motifs wherein the protein is associated with Lafora's disease.
2. A nucleic acid according to claim 1 having a sequence as shown in SEQ ID NO:1 (Figure 6A) or SEQ ID NO:3 (Figure 7).
- 10 3. An isolated nucleic acid molecule according to claim 1 comprising
  - (a) a nucleic acid sequence as shown in SEQ ID NO:1 (Figure 6A) or SEQ ID NO:3 (Figure 7A), wherein T can also be U;
  - (b) a nucleic acid sequence complementary to (a);
  - 15 (c) a nucleic acid sequence that has substantial sequence homology to a nucleic acid sequence of (a) or (b);
  - (d) a nucleic acid sequence that is an analog of a nucleic acid sequence of (a), (b) or (c); or
  - (e) a nucleic acid sequence that hybridizes to a nucleic acid  
20 sequence of (a), (b), (c) or (d) under stringent hybridization conditions.
4. A method of detecting Lafora's disease comprising detecting a mutation in a nucleic acid sequence according to any one of claims 1 to 3 in a sample from an animal.
- 25 5. A method according to claim 4 comprising detecting a C to G change at nucleotide number 205 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
6. A method according to claim 4 comprising detecting a T to A change at  
30 nucleotide number 76 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

7. A method according to claim 4 comprising detecting a deletion of nucleotides GA at nucleotide positions 1048 and 1049 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
- 5 8. A method according to claim 4 comprising detecting a deletion of nucleotides AG at nucleotide positions 468 and 469 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
9. A method according to claim 4 comprising detecting a deletion of  
10 nucleotide G at nucleotide number 992 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
10. A method according to claim 4 comprising detecting a deletion of 10 bp at nucleotide positions 373 to 382 in the *EPM2B* gene sequence shown in  
15 SEQ ID NO:1.
11. A method according to claim 4 comprising detecting a deletion of 32 bp at nucleotide positions 661 to 692 in the *EPM2B* gene sequence shown in  
20 SEQ ID NO:1.
12. A method according to claim 4 comprising detecting a T to C change at nucleotide number 260 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
13. A method according to claim 4 comprising detecting a A to C change at  
25 nucleotide number 905 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
14. A method according to claim 4 comprising detecting a T to C change at nucleotide number 98 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
- 30 15. A method according to claim 4 comprising detecting an insert of 2 Ts at nucleotide number 892 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

16. A method according to claim 4 comprising detecting a G to A change at nucleotide number 436 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
17. A method according to claim 4 comprising detecting a deletion of nucleotide T at nucleotide number 1100 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
18. A method according to claim 4 comprising detecting a deletion of nucleotide T at nucleotide position 606 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
19. A method according to claim 4 comprising detecting a A to T change at nucleotide number 923 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
20. A method according to claim 4 comprising detecting a G to T change at nucleotide number 580 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
21. A method according to claim 4 comprising detecting a G to T change at nucleotide number 199 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
22. A method according to claim 4 comprising detecting a G to A change at nucleotide number 838 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
23. A method according to claim 4 comprising detecting a C to T change at nucleotide number 676 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
24. A method according to claim 4 comprising detecting a deletion of nucleotide A at nucleotide position 468 in the *EPM2B* gene sequence shown in SEQ ID NO:1.

25. A method according to claim 4 comprising detecting a deletion of nucleotide C at nucleotide position 204 in the *EPM2B* gene sequence shown in SEQ ID NO:1.
- 5 26. A method according to claim 4 comprising detecting one or more mutations in the *EPM2B* gene as indicated in Table 1.
27. A method according to claim 4 comprising detecting a repeat of the sequence GCCGCCCCCGC (SEQ ID NO:5) at nucleotide position 1001 in  
10 the canine sequence of *EPM2B* shown in SEQ ID NO:3.
28. A method according to claim 22 comprising detecting at least 3 repeats of SEQ ID NO:5.
- 15 29. A method according to claim 22 comprising detecting at least 10 repeats of SEQ ID NO:5.
30. A method according to claim 22 comprising detecting from about 14 to about 26 repeats of SEQ ID NO:5.
- 20 31. A method according to any one of claims 4-26 wherein the animal is human.
32. A method according to any one of claims 4 and 26-30 wherein the  
25 animal is a canid.
33. A method according to any one of claims 4 and 26-30 wherein the animal is a dog.
- 30 34. An isolated protein containing a RING-finger domain and six NHL domains which protein is associated with Lafora's disease.

35. A protein according to claim 34 having the amino acid sequence as shown in SEQ ID NO:2 (Figure 6B) or SEQ ID NO:4 (Figure 7).
36. A method for detecting Lafora's disease comprising detecting a  
5 mutation in a protein according to any one of claims 34 or 35.
37. A method according to claim 36 comprising detecting a mutation in the *EPM2B* protein as indicated in Table 1.
- 10 38. A kit for carrying out the method of any one of claims 4 to 33 or 37 comprising reagents for the detection of a mutation in a nucleic acid sequence as shown in SEQ ID NO:1 or SEQ ID NO:3.
- 15 39. A kit for carrying out the method of any one of claims 36 or 37 comprising reagents for the detection of a mutation in a protein sequence as shown in SEQ ID NO:2 or SEQ ID NO:5.